Healthpoint

Information from the Division of Health Care Finance and Policy

Division of Health Care Finance and Policy

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Welcome to the 20th issue of *Healthpoint*! 20

The *Healthpoint* series started in 1996 as a quarterly issue brief for policy makers and has grown dramatically in popularity and readership. We welcome thoughtful suggestions that will ensure the value of future issues. To share your comments, suggest a topic, or request back issues, please contact the DHCFP Office of Communications: (617) 988-3125. Thank you for your interest.

GENETIC TESTING: ITS LIMITATIONS AND PROMISE

Until recently, genetics has played a relatively minor role in clinical medicine, primarily affecting the small segment of the population with a disease exclusively tied to a chromosomal abnormality or mutation. Genomics or the "new" genetics has uncovered genetic contributors to many common diseases and scientists predict that as more becomes known, genetic findings will inform numerous diagnostic, reproductive and therapeutic decisions. However, unlike the identified gene mutations that solely cause cystic fibrosis or sickle cell anemia, genomics uncovers predisposition, not inevitability, and this distinction has tremendous implications for how testing is used and how results are acted upon.

Scientists now believe that nine of the 10 leading causes of US mortality (injury excluded) will be shown to have some genetic component (see table below). Therefore, the political, ethical and social implications of genetic testing that have been the purview of relatively few families and their medical specialists will soon affect many of us and our primary care providers. This issue of *Healthpoint* examines genetic testing, its implications, as well as its limitations in predicting our individual medical futures.

The 10 Leading Causes of Mortality in the US

	Percent of Deaths in 1998
1. Heart Disease	32.0%
2. Cancer	23.2%
3. Stroke	6.8%
4. COPD	4.8%
5. Injury	4.2%
6. Pneumonia/Influenza	3.9%
7. Diabetes	2.8%
8. Suicide	1.3%
9. Kidney Disease	1.1%
10. Chronic Liver Disease	1.1%

Source: Alan E. Guttmacher, M.D., National Human Genome Research Institute, presented for APHA, November 14, 2000.

Genetic Tests

A genetic test is an "analysis of chromosomes, DNA, RNA, genes and/or gene products to determine whether an alteration is present that is causing or is likely to cause a specific disease or condition."[1] Genetic tests are recommended to determine definitively whether one has a disease, to diagnose the exact type of disease one has, or to determine the likelihood of one to develop or pass on a disease. The complexity of issues raised by testing leads most medical providers to recommend that the patient speak with a genetic counselor before and after testing. The genetic counselor helps evaluate risk and clarify treatment options especially amidst the uncertainty

of testing for predisposition. These sessions are not always covered by insurance despite the widespread belief that they are essential to informed and thoughtful decision making.

Genetic tests can be performed on a particular patient, a population (newborns), or those at risk for passing on a disease (a carrier). Predictive tests are performed on individuals determined to be at high-risk by their family history for developing a hereditary disease. These tests can be important for one found to have a genetic predisposition to a disease for which screening (i.e. mammography) or treatment exists. For example, for women who have a high preponderance of breast or ovarian cancer in their family, learning that they may have a genetic predisposition to the disease would alert them to the need for frequent screening. More aggressive interventions such as chemoprevention or even prophylactic surgery exist, but present painstaking choices to a patient who doesn't know whether she will in fact ever develop the disease. On the other hand, the vast majority of these cancers are thought to have other causes (smoking, poor diet, exposure to carcinogens, etc.), so one couldn't take complete comfort from a negative genetic test or abandon healthy living or regular screenings.

Although the popular media's focus has been on predictive testing, experts estimate that it is diagnostic testing that accounts for 90% of all genetic tests currently performed. These tests diagnose a genetically related disease in a person exhibiting particular symptoms or they identify the genetic makeup of one's previously identified condition to target a treatment regimen. Insurance coverage for predictive tests varies while diagnostic tests are generally covered. Many insurers have no written policy on genetic testing, determining coverage on a case-by-case basis.

Testing to Inform Reproductive Decisions

Carrier testing is performed for family planning purposes prior to conception to determine whether an individual carries (and therefore may pass on) one copy of an altered gene for a particular disease. Tay-Sachs carrier testing, begun in the 1970s among the higher-risk eastern European Jewish population, has proven successful in significantly reducing the number of Jewish babies with this disease conceived by two carriers, thus lowering the incidence of this fatal disease. Other diseases also occur in specific ethnic populations at higher frequency than in the general population, so primary care physicians should suggest and make available carrier testing when applicable.

Since there is no carrier test (which would be preferable) for many birth disorders, prenatal diagnostic genetic testing is recommended for those at high risk. Amniocentesis, approved by the Federal Drug Administration (FDA) in 1967, is one procedure used especially in pregnant women over age 35 to test for a variety of abnormalities, most famously Down's syndrome. When a test reveals a significant anomaly, parents face an emotional decision—to terminate the pregnancy or to prepare for the birth of a child with special health care needs. As a result of prenatal screening, amniocentesis and subsequent pregnancy termination, the number of babies with significant anomalies born to older mothers has decreased.

Newborn Screening

Each state requires its newborns to be tested for some number of genetic disorders, making newborn screening the most common application of genetic testing today. Massachusetts' newborn screening program, established in 1962, is one of the most comprehensive in the country, screening newborns for 10 disorders plus currently, an additional twenty through an optional pilot program. For a disorder to be included in the required screening program, the test for it must be reliable and

the disorder treatable through early medical intervention. Screening at birth is crucial for the small group of babies with rare diseases for whom immediate treatment prevents mental retardation, disability or even death.

The Massachusetts Genetics Program

In 1980, Massachusetts became the first state to recognize the increasing importance of genetics with the establishment of the Massachusetts Genetics Program within the Department of Public Health. The Program has been responsible for facilitating the development, delivery, and appropriate utilization of high-quality, family-centered, community-based, culturally and linguistically appropriate genetics services and public policies. Today, the Program is conducting a statewide genetics needs assessment and strategic planning process in order to plan for its expanded role as the field of genetics extends its reach to many more of us.

Insurance Discrimination and Privacy

As the number of genetic tests available increases and one's disease predispositions become more accurately identified, there is heightened concern about potential discrimination in employment and insurance decisions. State laws on genetic information privacy and discrimination were first enacted in the 1970s, but these laws were disease-specific and thus narrow reaching. Many states have enacted broader protections; Massachusetts became the most recent state to do so in August 2000. This new law bans insurance and employment discrimination based on genetic information and includes provisions on privacy and informed consent prior to testing. Today, thirty-three states prohibit discrimination for health insurance based on hereditary disease factors, twenty states require the insured's consent to disclose genetic information and 22 states prohibit employment discrimination based on genetic information.[2]

In addition to state-level activity in the genetics arena, federal legislative action has been taken to ensure a national standard of protection and to reach the self-insured exempted from state law. The 1996 Health Insurance Portability and Accountability Act prohibits an insurer administering a group plan from using genetic information to deny coverage or increase premiums. In addition, the Secretary's Advisory Committee on Genetic Testing was chartered in 1998 to advise the Secretary of Health and Human Services on issues raised by the development and use of genetic tests.

Policy Concerns

In recent years, many commercial developers of genetic tests have patented both the tests and the genes upon which they are performed, sometimes charging royalty fees per test and requiring facilities to pay a fee for designation as a testing site. The biomedical industry insists that the practice of patenting is fundamental to promote and protect research and development. Opponents of gene patenting counter that this will encourage companies to either prematurely introduce genetic tests to recoup investment more quickly or inappropriately broaden the definition of an at-risk population in an effort to increase its utilization. For example, the company which holds the patent for the breast cancer genetic test doesn't exclude from its testing criteria women without a family history of breast or ovarian cancer despite widespread medical agreement that it is women with a high prevalence of those diseases in their family who are most likely to have a genetically linked cancer.[3]

Another area of policy concern is that some testing facilities are already advertising directly to consumers, a strategy mimicking the pharmaceutical industry. Since 1997 when the FDA relaxed

its advertising rules, there has been a huge increase in pharmaceutical advertising and subsequent consumer demand for name-brand drugs. Direct to consumer advertising for gene testing might dramatically increase our medical costs if those who seek it are primarily the "worried well" rather than those at high risk. Therefore, policy makers should strongly consider prohibiting the genetic testing industry from following the same unfiltered path to consumers that the pharmaceutical industry now enjoys.

The Future of Genomics

The rapid pace of genetic discovery has been driven in part by the 1990 launching of the Human Genome Project, an international public and private collaborative effort to identify the approximately 100,000 genes in human DNA. It is anticipated that its findings will not only increase the number of genetic tests available but lead "to a new era of molecular medicine characterized less by treating symptoms and more by looking to the most fundamental causes of disease."[4] The Project is expected to spur advancements in the emerging field of pharmacogenomics, "the study of how an individual's genetic inheritance affects the body's response to drugs."[5] It is anticipated that physicians will be able to prescribe drugs designed to work for an individual's specific gene mutation, thereby leading to a more efficient system of prescribing with fewer adverse drug reactions. While this application may be many years off, there is no doubt that in the meantime the impact of genetics will continue to grow in all areas of clinical medicine.

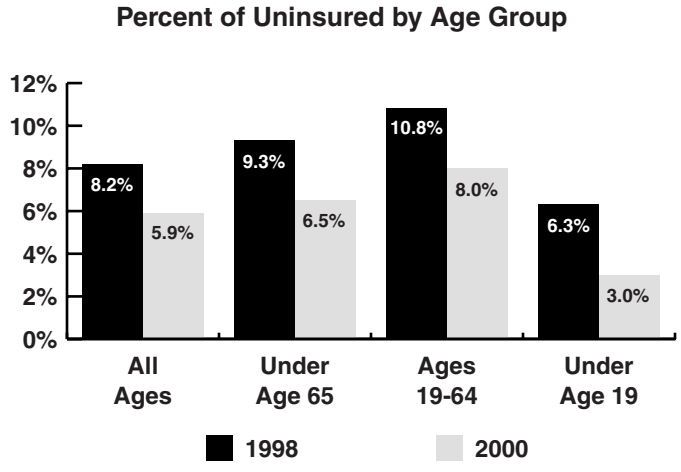
Endnotes

1. Federal Register, Vol. 65, No. 76, April 19, 2000, Part IV, Department of Health and Human Services: Secretary's Advisory Committee on Genetic Testing; Notice of Meeting.
2. National Conference of State Legislatures, LegisBrief, June/July 2000, Vol. 8, No. 28, "Protecting Genetic Information" by Cheye Calvo and Jennifer Jones.
3. Clinical Genetics, Vol. 57, No. 5, May 2000, "Genetic Testing, Ethical Concerns, and the Role of Patent Law" by Timothy A. Caulfield and E. Richard Gold.
4. Human Genome Project Information, "Potential Benefits of Human Genome Project Research" web site: http://genome.rtc.riken.go.jp/hgmis/project/benefits.html
5. Human Genome Project Information, "Pharmacogenomics: Medicine and the New Genetics" web site: http://genome.rtc.riken.go.jp/hgmis/medicine/pharma.html

Did you know?

Fewer Individuals in Massachusetts Are Uninsured in 2000

In 1998, the Division of Health Care Finance and Policy initiated a statewide survey of the health insurance status of state residents, and repeated it in 2000. The results indicate that approximately 9.3% and 6.5% respectively of all non-elderly Massachusetts residents were uninsured at the time of the interviews. In both years, the largest group of uninsured individuals were ages 19-64. Those under age 19 showed the greatest rate of decrease between 1998 and 2000, most likely due to the State Children's Health Insurance Program, S-CHIP, a federal expansion inspired by a Massachusetts reform, Chapter 203 of 1996.



Source: Division of Health Care Finance and Policy Year 1998 (Area Probability Sample) and Year 2000 Health Insurance Survey Results

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